



Rare disease curation takes minutes instead of weeks via automation using genetic and medical knowledge with proven algorithms

CENTOGENE implements ACTICO's platform to dramatically accelerate and improve the quality of diagnostic data curation workflow.



Success Story

Objective

Accelerate rare disease diagnosis and research, by automating the manual curation of diagnostic data for CENTOGENE's proprietary CentoMD® database.

Implementation

Automation of the data curation process via the ACTICO Business Rules Engine, implemented in partnership with Business Rules specialists S&D.

Results

Data curation process fully automated, reducing results delivery period from one month to five minutes.

RARE DISEASE DIAGNOSES IN MINUTES, NOT WEEKS

For patients who are suffering from an undiagnosed rare genetic disease, receiving a diagnosis as early as possible is vital. As a world leader in genetic diagnostics for rare diseases, Rostock-founded CENTOGENE GmbH is committed to accelerating the diagnosis of these conditions. Thanks to their partnership with Business Rules specialists S&D, and the implementation of the ACTICO decision management platform, the company has recently made significant progress in that endeavor.

A UNIQUE GENETIC DATABASE

Since its inception in 2006, CENTOGENE has developed a unique genetic information database, CentoMD®. CentoMD® 5.8 currently contains over 430,000 analyzed cases as of August 2020 and is a powerful tool in making the rare disease diagnostic process easier, faster, and more precise. CENTOGENE's Director of Artificial Intelligence, Prof. Dr. Carsten Ullrich, explains, "Our goal is to accelerate the diagnosis of rare hereditary diseases with a genetic cause. We receive thousands of blood samples from around the world from physicians using our CentoCard® solution. This innovative filter card



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Prof. Dr. Carsten Ullrich, Director of Artificial Intelligence, CENTOGENE

uses Dried Blood Spot (DBS) technology, which makes sending patient samples as easy as sending a letter. From each DBS, we are then able to generate genetic, enzymatic, and biomarker profiles of the patients, which our medical experts evaluate to produce a diagnosis within days. It's important to remember that because it's often difficult for the medical community to identify rare hereditary diseases, it often takes up to eight years for a correct diagnosis to be made. That's why the ability to speed up the diagnostic process, thereby enabling patients earlier access to treatment, is so crucial. It can help avoid a lot of patient suffering. We already have a very well established bioinformatic pipeline that allows us to analyze genetic data and identify the mutations of an individual patient that cause their rare genetic diseases."

ACCELERATING DIAGNOSTIC DATA CURATION

Curation of key diagnostic data has always involved manual processing that could be very time-intensive. This is particularly challenging when, for example, new information about genetic mutations becomes available, and needs to be integrated into the database. Prof. Dr. Ullrich explains, "We have a range of standard operating procedures that our medical professionals complete for data curation, and we were exploring the time saving opportunities that artificial intelligence automation solutions could offer. This process provided an ideal test case for us to find out what is possible, how easy it would be to integrate AI into the workflow, and how flexible it would be."

FINDING THE RIGHT PARTNERS

In early 2019, CENTOGENE was looking into using their rules-based decision-making engine for automating the curation process. "I was immediately impressed with the expertise of S&D, and their willingness to explain the pros and cons of the various Business Rules platforms objectively," Prof. Dr. Ullrich explains.

During their conversations, the S&D team suggested ACTICO as the most suitable platform for automating CENTOGENE's data curation process. "The user interface and graphic representation of the decision-making process was very clear, which closely resembles the general decision-based system that our staff had been using. The ease of implementation and predictable costs were also very attractive. In the end, it was an easy decision. S&D experts were able to get up to speed with our requirements very quickly, and they have remained extremely responsive to our needs. The ACTICO solution proved to be simple to integrate with CentoMD®," adds Prof. Dr. Ullrich.

Of course, regardless of how straight forward the integration is, it's always important to build user trust in a new system. That's why CENTOGENE decided to take a two-stage approach to implementation:

1. Integration of the system into the user workflow – allowing users to check that the results being produced by the ACTICO platform made sense.
2. Increasing levels of automation until all manual processing tasks were eliminated

Prof. Dr. Ullrich says, "Importantly, this approach enabled us to ensure great transparency. We were able to discuss the decision-making rules with domain experts as they were implemented within the ACTICO platform. This made it easy for them to verify that they had been implemented correctly."



A BREAKTHROUGH IN SUPPORTING PATIENTS

Dr. Martin Werber, Senior Technical Project Manager for AI at CENTOGENE, explains the value for patients that has been achieved through the collaboration with S&D and the ACTICO platform, “We are now able to batch the processing of thousands of patient samples with this fully integrated solution and save up to one month of work for every case. For patients, this represents a huge breakthrough. The time previously spent looking at less complex cases each month can now be spent studying more of the most difficult – meaning patients directly benefit.”

He continues, “Our quality control process can be easily applied retrospectively, to identify cases where new information has become available that enables a diagnosis. This is crucial in terms of the lifetime commitment we make to our patients. If a negative test result is reclassified over time as the result of new information, we inform the patient about the possible change in diagnosis. The automated curation process we have established with S&D and ACTICO helps us keep this promise even more efficiently.”

Prof. Dr. Ullrich adds, “The automation we have implemented further proves our position as a technology leader, because it increases the quality of our CentoMD® database by minimizing human intervention, and ultimately helps us deliver better, faster diagnoses. **This is possible because the curation process that used to take a month now takes five minutes. It’s dramatic proof of how the ACTICO platform gives us the confidence to apply a similar approach to other processes in the future. Everyone trusts it.**”

Looking to the future, both Prof. Dr. Ullrich and Dr. Werber see the opportunity to replicate this success with other similarly manual, but essential processes, “We can see a future in which we make use of the Machine Learning module within the ACTICO platform. We are very much looking forward to exploring what else we can do with the technology, supported by S&D,” concludes Prof. Dr. Ullrich.



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Dr. Martin Weber, Senior Technical Project Manager for AI, CENTOGENE

CENTOGENE

CENTOGENE engages in diagnosis and research around rare diseases transforming real-world clinical and genetic data into actionable information for patients, physicians, and pharmaceutical companies. Our goal is to bring rationality to treatment decisions and to accelerate the development of new orphan drugs by using our extensive rare disease knowledge, including epidemiological and clinical data, as well as innovative biomarkers. CENTOGENE has developed a global proprietary rare disease platform based on our real-world data repository with over 3.6 billion weighted data points from approximately 570,000 patients representing over 120 different countries as of August 31, 2020.

The Company's platform includes epidemiologic, phenotypic, and genetic data that reflects a global population, and also a biobank of these patients' blood samples. CENTOGENE believes this represents the only platform that comprehensively analyzes multi-level data to improve the understanding of rare hereditary diseases, which can aid in the identification of patients and improve our pharmaceutical partners' ability to bring orphan drugs to the market. As of August 31, 2020, the Company collaborated with over 40 pharmaceutical partners covering over 45 different rare diseases.



ACTICO is a leading international provider of software for intelligent automation and digital decisioning. Its scalable software combines rule-based technology and machine learning in a unique way and is totally audit-proof. It allows companies of all sizes to process huge amounts of data and make and automate AI-supported and rule-based decisions in real time. Through intelligent automation, ACTICO increases the business value of its customers by improving their operational decisions.

Today, its customers include leading companies in 25+ countries ranging from small/mid-size to Fortune 500 companies. ACTICO has offices in Chicago and Singapore and its head office is located in Immenstaad, Germany.

More information at www.actico.com

ACTICO

Europe

ACTICO GmbH
Ziegelei 5
88090 Immenstaad
Germany

Americas

ACTICO Corp.
141 W. Jackson Blvd.
Ste 300A
Chicago, IL 60604
USA

Asia & Pacific

ACTICO Pte. Ltd.
#11 - 04, The Arcade
11 Collyer Quay
049317 Singapore

info@actico.com
www.actico.com

